

Case conference

Limits of confidentiality¹

A patient is described, a woman of 60, who at that age was diagnosed as having Huntington's chorea, a genetic disease which is transmitted as an autosomal dominant². She had one daughter, who was married and lived abroad. The patient's mother and maternal aunt, it emerged during the consultations, had had the disease, and shortly after the diagnosis had been made the patient's sister was admitted to hospital with a respiratory condition, and in the course of treatment Huntington's chorea was diagnosed in her also. She had one daughter. Throughout, the problem was not only that of treating the patient herself, but the situation of all those of the family affected in any way. The patient's husband had not known of her medical history at the time of the marriage. At no time had the daughter been told.

The discussion, in which seven doctors, a moral philosopher, and a social worker took part, centred on the basic issue of confidentiality in the case of a disease, from which would stem problems for the affected family through several generations. All were agreed that in these circumstances those affected must be told, but differed as to who should make some of the subsequent decisions. Some thought that having told of the diagnosis and its implications, the doctor should leave further decisions to those concerned; others that in a disease such as Huntington's chorea one is looking at part of a whole chain of genetic material, and, although the individual patient or potential patient is of course important, he or she must be seen in the genetic setting of the community. The participants also considered the psychological

support that would be required for affected individuals and families. No one advocated compulsory sterilization for these patients or carriers.

Case history

A 60-year-old married woman was referred to a general psychiatric outpatient clinic by her general practitioner for advice on the treatment of depressive symptoms which had been present for about six months.

The patient had married a Danish man some years younger than herself when she was in her mid-30s. The only daughter of the marriage had lived with her parents for most of her life. When she became pregnant in her early 20s, she had remained at home and the child, a son, had been brought up largely by his grandparents, while his mother went out to work. The grandparents had become very attached to this boy and his grandfather had begun to teach him Danish. The daughter then met another man who wished to marry her but was unwilling to adopt her son. Eventually the son was adopted, through an adoption society, and the daughter married, the couple emigrating to a former British colony.

The patient and her husband were greatly disturbed by the loss of the grandson, although pleased by the daughter's marriage, and confident that the grandson was being well cared for. The patient felt low spirited and her mind was filled by thoughts of the boy. There was a clear link between recent events and the patient's symptoms and she was regarded as suffering from a pathological mourning reaction to the loss of the boy. Treatment was directed to improving her mood and helping her to come to terms with her loss.

After the patient had been attending the clinic for about six months, she began to complain of unsteadiness in walking. At first it seemed that this might be due to the sedative effects of medication, but it soon became clear that not only was her gait unsteady but that there were choreiform movements of the hands and grimacing movements of the face. These movements had been disguised by the patient in a variety of ways.

Arrangements were made for the patient to be admitted to hospital for observation. Until this time there had been no hint of any family history of mental illness, but shortly after admission the patient revealed that her mother and her maternal

¹Those taking part in this case conference were: Miss Margaret Atkin, principal medical social worker, The Hospital for Sick Children, Great Ormond Street, London; Dr Alastair Campbell, Editor of the Journal; Dr Leslie Cann, psychiatrist, Nottingham; Dr Adrian Caro, general practitioner and chairman of COMBAT; Dr Roger Higgs, general practitioner, London; Professor Marsden, Institute of Neurology, Maudsley Hospital, London; Dr Raymond Pietroni, general practitioner, London; Dr Will, lecturer in medicine, Nottingham Medical School; Dr John, the consultant psychiatrist presenting the case.

²This means that Huntington's chorea is passed dominantly, that is, on average to half of the affected parent's offspring irrespective of sex.

aunt had had movements of the face and limbs, beginning in their 40s and continuing until death, and that her living sister also had similar movements. The aunt had been an inpatient in the same hospital and her notes gave a diagnosis of Huntington's chorea. The patient had long suspected that she had the same disease as other members of the family, but when this was confirmed she forbade the doctor to discuss it with her husband, who had never been informed of the existence of an hereditary disease in his wife's family; or to inform her daughter, who was in any case out of the country. The patient was of sound mind and this was a powerful injunction. The psychiatrist was anxious that the daughter be informed of the danger of her transmitting the condition to further children. The best chance of informing her was through the father, the patient's husband, but the patient would not countenance his being told. This ethical dilemma was not resolved immediately, but there were grounds for hope that the patient would change her mind and the matter was allowed temporarily to rest.

A few months later the psychiatrist had a telephone call from a house physician at the local district general hospital. He had recently admitted the patient's sister for a respiratory condition. The sister showed unmistakable chorea, confirming our expectations that she too was suffering from Huntington's chorea. It was subsequently agreed by all medical staff concerned that the psychiatrist should assume the central role in counselling and treating the family. Shortly afterwards, the patient's niece, the daughter of her affected sibling, unexpectedly came to the outpatient clinic to see the psychiatrist as she was concerned about the health of the patient, her aunt, who was due to visit the clinic, accompanied by her husband, that morning. No change had occurred in the patient's attitude but the niece was now clearly involved. The psychiatrist told the patient's niece that both her aunt and her mother were suffering from the same condition, and that this was hereditary. It was appreciated that this was strictly a breach of confidence, but the inference would probably soon have been made by the family. Arrangements were made to see all of the sister's family to discuss the illness with them. Later that morning the psychiatrist told the patient that the niece had been informed and that she intended to tell her husband of the hereditary nature of her illness. When the husband was informed, it was clear that he had already suspected that the illnesses in the family were linked but had not realized that his daughter and her offspring might be involved. Not unnaturally, he was angry at what he saw was a serious deception by his wife's family. The husband agreed that the psychiatrist should write a brief letter of explanation for him to send to his daughter.

After consultation with the affected sister's medical attendants, the psychiatrist and a social

worker saw her family, comprising the brother-in-law, the niece with her husband and two children, the nephew, with his wife and two children. They were informed of the general nature of Huntington's chorea, and the risks of its being inherited by other members of the family. It was stressed that there is, as yet, no certain way of detecting carriers of the disorder before they show evidence of the disease, and that they could not assume themselves to be free of the risk of both developing the condition and passing it on to their children. This aspect of the condition was reinforced by the late onset of the disease in several members of this family. The younger members of the family were advised that it would be unwise for them to have further children.

A few weeks later the situation was complicated by the niece becoming pregnant. She decided to seek an abortion, but her request was not well received as the gynaecologist did not appear to understand the mode of transmission of Huntington's chorea. Following this rebuff, the niece's family were referred by her general practitioner to the local genetic counselling service. The information they had already received was confirmed by this second opinion, and the abortion was subsequently carried out.

The patient's sister was referred to the psychiatrist; this interview simply served to confirm the diagnosis and allowed the family history to be checked from another source. It was evident that the sister had suffered some impairment of memory.

The present situation with regard to this family may be summarized as follows. It has been confirmed that the family suffer from Huntington's chorea; many of the relatives have been informed and the nature of the disorder has been explained to them; the family have had expert genetic advice from an independent source; one member of the family has had a termination of pregnancy on genetic grounds. Further, it is almost certain that the patient's daughter has been informed of the disease, although it is less certain that her husband has been informed, or that she fully understands the implications of the disease. As so many of the family now know of the disease, the patient's daughter is likely to be persuaded to visit the psychiatrist when she is in Britain at some future date.

This history raised a variety of ethical problems but the central issue was the inclination to place the wider implications for the family above the patient-doctor confidence. It was believed that the husband was entitled to know something of the illness, even though it was against the patient's wish, as it could affect his daughter as well as his wife. Further, it was thought important that other members of the family should not continue to bear children in ignorance. This of necessity involved more distant relatives, who were seen without

request from their own practitioners and without the consent of the patient. In the event, it was possible to moderate the breach of confidence, information being given as the family became aware of the connexion between the patient's and her sister's symptoms.

Divulging medical information

DR CAMPBELL

Perhaps we could start our discussion on the question of divulging information to the husband and other members of the patient's family. Is this ethically justified?

DR CARO

Well, the information isn't the property of the patient – it is the property of the family. They are carrying the gene whether they know it or not and have this particular potentiality, and it's their property whether they know it or not. Whether you *tell* them is another problem.

DR JOHN

But in your view, did the patient's husband have a *right* to know that his wife was carrying, or rather, had this disease?

DR CARO

In my view he does. A number of patients' spouses whom I have spoken to, who had discovered the facts after marriage, felt that they had been cheated in some way and were very, very angry about it.

DR JOHN

This was true of both of these husbands.

PROFESSOR MARSDEN

It is difficult when sorting through these problems. One has to think about precedents outside Huntington's chorea before divulging medical information to members of a family about an individual patient. The one that always comes to my mind is the question of syphilis. When one is faced with a patient with an infective disease of the nature of syphilis one has no hesitation whatsoever, whatever the patient says, in contacting or tracing those husbands and children and other contacts. We know full well that these are at risk of developing disastrous illness unless caught and treated.

Here, I think, one does dismiss the question of patient confidentiality for the sake of other patients who become one's responsibility through one's individual patient. And I think this is a guiding line in this particular case, too: one has a responsibility to other people.

DR CARO

Yes, you can break a rule of 'medical ethics', possibly with the excuse that it is in the best interests of the patient. There is also society's problem. Do you stop people contriving to protect themselves or society?

DR WILL

It is very easy, of course, to get the analogy slightly wrong. Indeed I think there are no exact analogies. Syphilis, for example, is likely to go beyond the contacts of the person concerned: so that, although you obviously have got a continuation of the chain, it will go beyond a family. You can draw three groups: there is the individual; the family – in Huntington's I would say very important; and society – in Huntington's perhaps much less important than in the case of syphilis. So perhaps this isn't an exact parallel. Secondly with syphilis, information is treatment, and information is treatment is cure. That again, is not quite the parallel here, is it?

PROFESSOR MARSDEN

The analogy is not quite as you put it. I don't think the fact that it is a community, rather than a family, problem is relevant to the argument. The critical thing is, Are you breaking the confidentiality of the individual patient, either to the family or to the community? Whether it is the family or the community I don't think it really matters in that argument.

Why one is doing that in the case of syphilis is to inform the contact of the risk they are taking and the measures that are necessary to prevent it causing trouble. That is exactly what one is doing in Huntington's chorea, to my mind.

DR PIETRONI

I don't really feel that the analogy holds. This woman had finished her childbearing and therefore nobody was at risk from her having this condition. It seems to me easier to look at it in a different way. When you know she has Huntington's chorea she's telling you two things: one, that she has a condition, and that is her condition and it is her information, and it's up to her to whom that is divulged; but she's also telling you something about her family. You're thus being told something willy nilly about the daughter, and it would seem that the patient has no right to insist that you keep the information from her. I can see that you could keep it from the husband because, as you say, her childbearing was finished, but presumably it would be perfectly ethical to inform the daughter in some roundabout way that this information had been discovered. But I don't really see that one has a right to tell the husband against the patient's wishes.

MISS ATKIN

When I read your case history I was interested in why this woman didn't want the rest of the family to be involved. Was there guilt? Were there other problems in the marriage? Other deceptions? It's a very angry and rather bitter person who doesn't mind (from what she knew of the condition) that there was going to be a lot of suffering for other members of the family or who was not aware what

this would mean in terms of her relationship with everybody.

DR JOHN

Well she certainly wasn't getting on with her husband. There were several factors. One was that the age gap between them was becoming increasingly apparent. The husband was drinking and she complained to me about this. He also had gall-bladder disease and had been advised to have an operation which he couldn't face up to. So he was really quite unwell at the time himself.

DR CARO

If husbands or wives are told, in the main, once the unaffected spouse is aware that the problem is not their fault but that there is something causing it, in my limited experience, they will stick together. By telling the unaffected spouse that there is a reason for the patient's strange behaviour, the help that he or she is then prepared to give to the affected is increased because of the understanding. So it is important to tell in the patient's interest.

DR JOHN

One of the reasons why she was very sensitive about it was that it wasn't the only deception. I think there had been some deception about her age at the time of marriage as well.

PROFESSOR MARSDEN

I think you've highlighted a very important point in this. Are you, when dealing with something like Huntington's chorea, going to serve their best comfort of mind or social circumstances by telling them lies? Or are you going to tell them the truth and face the risk of utter disaster happening as a result of it?

From what you've just said, it may be better to respect that lady's privacy, preserve her marriage, and risk all the children that come from the whole line that she has created.

DR PIETRONI

It seems to me a very important principle that you cannot protect people from important facts about their lives. If they commit suicide, that is not a case for not telling them; it is a case, perhaps, for telling better. Inevitably, I agree, there will be some who will react badly but it's a very dangerous precedent to keep back information, important information, on the basis that they won't be able to cope with it.

DR JOHN

Although none of this family has committed suicide it has certainly disturbed them very considerably. More than amongst the people of the same generation as the two patients, the disturbance has been in the people of the next generation. They have learned from me and from the paediatricians that they themselves *might* have the disease but we

can't tell at the moment, and that their children *might* have it but we can't tell at the moment.

Identifying those at risk

PROFESSOR MARSDEN

This provokes another ethical problem. About 70 per cent or more of patients develop the disease after the age of 35 to 40. You make a diagnosis on a 35-to-60-year-old who has undoubtedly got the disease. The whole family history then becomes apparent. You are thus left with a number of children of the person who has the disease, none of whom show the disease because they are all in their 20s. They all then come to you saying, 'I am pregnant', or I want to get pregnant. I haven't got the disease now. What should I do about it?

All you can tell them at that age – they are now 20 with no physical signs and completely normal – is that each child of theirs has a risk of 1 in 4 of developing the disease.

DR CARO

Although one looks at the individual in terms of the ethics related to an individual, if you look into the Huntington's gene it is part of a whole chain of the same genetic material that you cannot possibly look at in isolation; it's a genetic disease. You can't look at one person and say, 'That's it', and shut your eyes to other sides of it.

PROFESSOR MARSDEN

This is where you and I part company! I think personally that I have an absolute duty to potential carriers of Huntington's chorea to give them the facts on which they could then make their own decision. I do not see personally that the doctor's role is to control progressive disease in society. If I did, then a whole series of events would follow. One would be that one would break every ethical rule one could think of to trace sufferers from Huntington's chorea all over the country, to get hold of them. The next one would be to sterilize everybody who's got Huntington's chorea and abort every child of a potential sufferer of Huntington's chorea, in order to stamp out the disease. That, to my mind, would be the logical conclusion of trying to eradicate the disease; and I think that infringes so much upon the individual's right to decide for themselves what they want to do – given the correct information – that I don't think it's the right thing to do.

DR CARO

I'd like to put my 'half' of where we divide company, as it were. What I would like to see is that anybody with a family history of Huntington's has it noted on the general practitioner's records. I'm not suggesting you should sterilize them: but if biochemists came up with a method of finding a marker, or a treatment, then we've got to know who are the people at risk. There's no point in looking

for a biochemical answer to this disease unless we know to whom to give it if it is found. It's presumably got to be given before the brain is damaged. So you have got to know who these people are if any help is going to be given to them later on. And this does mean looking for them.

Why inform those at risk?

DR PIETRONI

Why should we have to identify the sufferers before we have something to offer them?

DR CARO

If you look at it before they have the disease, early on, it does make a difference if you notice that they are potentially carrying the gene. There are a number of problems present earlier on, and if you can identify these problems you could stop diagnostic dilemmas arising – if you know the family history. For instance – somebody, say, who had been charged with rape – you could say you were protecting society by putting him in prison. But he shouldn't have been in prison – he should have been in a mental hospital.

DR WILL

I find this a rather tenuous line to follow. You may destroy a person's ability to provide for their family because, once having told them that they have this kind of condition gratuitously, situations like insurance become impossible for them.

DR CARO

The point is to assist a family *before* the diagnosis is made, or before they come to the psychiatric hospital, before they go to the neurologist. You could see work failing, people being on the dole, for reasons that are not evident. You could say to the social service workers, 'This isn't just a passenger'; and you could say to the wife, 'Do you realize what's going on? We'll see how we can support him'. You can do all sorts of things, get housing changed, before the diagnosis is made, if you can make it early; and you can only make it early if you are aware of the possibility.

DR HIGGS

But this ignores the way in which human behaviour occurs. You are very much looking at life biochemically, as it were. One meets several families where somebody has decided that they are ill and they go off and cease to work properly, when there is, in fact, nothing wrong with them at all. So that you may well, by so-called 'helping' that family, put a burden on them which may make them – through depression, or anxiety, or whatever psychiatric label you give it – just give up work and do just the things that you describe as being the signs of early Huntington's. It worries me that anticipated disease, even if not present, can actually become expressed. One has seen children of

epileptic patients who may then develop epilepsy with normal EEGs and one doesn't know what it is – is it hysterical or what? I don't know whether people have developed hysterical Huntington's, but it seems to me that the stage would be set for just this sort of reaction if we went along and informed every single young person that they were liable to the disease.

PROFESSOR MARSDEN

The moment you tell an unaffected person at risk you can be certain that 50 per cent of these people are going to be back over the next few years saying, 'I've got a twitch over my left eye; is it the disease, doctor?' That happens time and time again. The situation may be made even more difficult by the fact that in the next decade there will most probably be a predictive test for Huntington's chorea. You could then take 20-year-olds and give them something or measure something which would enable you to tell whether they would develop the disease or not. Fifty per cent of the people you give the test to will be negative; and if the test is right you could go ahead and tell them they could have children as they don't have the disease and won't get it. To the other 50 per cent you would have to say that they would reproduce the disease.

DR JOHN

The news that there is Huntington's chorea in the family can't be wrapped up in a pleasant package. It must be presented fully.

DR WILL

Are you telling them in such a way as to stop them having children?

DR JOHN

Absolutely: there's no point otherwise!

DR WILL

But this is at odds with what we have said. It was said that this was the patient's decision. Feeling as you do you could never tell them in a neutral fashion: you must project what you feel, and certainly some part of what you feel is likely to come over without you knowing it. One cannot be neutral but should one, in fact, be quite so aggressive about saying, 'Yes, this is my intention: I do want to stop them having children'?

PROFESSOR MARSDEN

It depends entirely on what your attitude is – what you want as they walk out of the door. When they walk out of the door I want them to really know what the facts are about Huntington's chorea. You can't wrap that up in any way whatsoever. But they need to know the true facts in order to make their own decisions about their life and family.

DR PIETRONI

I don't agree with you. If I were faced with a patient and I had to tell them, I wouldn't know

whether they should have children or not. Therefore, my feelings would be neutral. I wouldn't know: I would like to help them come to a decision – in the same way with a request for a termination of pregnancy. You may have very strong feelings, but in general I don't know what's right and what's best. I do know that I have facts that I want them to consider, and I want to help them to come to what is the right decision for them. I think that if you do have strong feelings, then perhaps you shouldn't be doing that particular counselling.

DR CARO

What I say to such patients is, 'My advice to you is perhaps not to have children for five years.' I don't even suggest they should get sterilized. I say, 'We don't know what the situation in predictive testing is going to be. We don't know whether we're going to know. We don't know whether there's going to be treatment. But don't have children for five years, and then come back and ask me.'

DR PIETRONI

With respect I suggest that we should look very carefully at how we set about these things; and perhaps non-medical people could help us in this matter. You say about telling them and that sometimes it doesn't get across. I don't know how many times you see them but, as a general rule, the first reaction to something unpalatable is denial, switching off; and it may be months before they go through the anger stage and depression stage – such a patient may through bereavement or facing death – finally get to the stage of acceptance, which many people never reach. Perhaps Miss Atkin could help us here.

MISS ATKIN

I see very few patients with Huntington's chorea and I don't have the experience of this particular condition that the rest of you have, but most of my work is in facing families who are bearing the loss of one child after another with genetically determined diseases.

I am usually with the consultant when he sees people. There are always two interviews because one usually feels that the qualitative pace and level at which parents hear things is different: what one assimilates, the other one doesn't: and often, however much they may be integrated as a couple and at one in many things, they aren't at one in hearing this sort of thing. So, invariably, there are at least two interviews, and I think very few of them leave the hospital with any doubt as to the implications of the condition that their child has. It is my task, usually, to pick up the pieces after this sort of interview and help them with their initial reactions. Usually they are asking for specific advice, and one is helping them to make decisions with as much awareness of themselves, their own psychology, the quality of their marriage, and everything else,

as to what is right for them rather than what they should do about future pregnancies.

Parents have lots of problems in communicating information to their older children. One also finds with many of these families that their priorities and philosophy and everything else change profoundly. Their expectation of life and of their children, and of society, and of care givers, is measured somewhat differently as a result of what they've experienced.

DR HIGGS

Is this in any way an enriching thing, sometimes?

MISS ATKIN

Yes, sometimes. Certainly, it isn't with everybody, but it can be. Sometimes it is a very negative one: and one is in the situation of the parents invariably asking – as I'm sure they do with all of us, 'Which of us is to blame?' and 'Whose genes are they?' I try, if I can, to meet the parents in the prediagnostic phase so that one has got a bit of an idea, even if it's only a thumbnail sketch, of what sort of marriage, what sort of family, what sort of stresses there are before they learn about the one that's coming at the end of the week: before you precipitate another crisis with the bad news, what sort of inner resources they have got, what sort of people they are, what sort of communication goes on in the family, and what sort of capacity they have to cope with what they are going to hear.

DR LESLIE CANN

Isn't it by helping people individually that the solution is found to the doctor's problem of confidentiality in the case we have been discussing? If you are trying to answer the patient's original query – which was that she was depressed – then you will work with her guilt; and with her problems of being ill. That will lead on to the problem of her having to tell the rest of the family, and that responsibility can remain hers. You can help her face it but not take it upon yourself as a doctor to impose your solution on her.

DR CAMPBELL

Here I think we must end the discussion. We can't claim to have 'solved' any of the problems. Like most difficult decisions on medicine, there are no easy solutions. But, if I may attempt to summarize a very complex discussion, we seem to be generally agreed that drastic measures to eradicate the disease (such as compulsory sterilization) are not ethically justified; that information about its prevalence in a family should be communicated – especially to those of childbearing age – even if this entails some breach of confidentiality; and that the skill and sensitivity with which the information is communicated is a central moral aspect of the problems which the case presents. Even if we cannot cure the disease, we must try to safeguard the integrity of those whom it threatens.